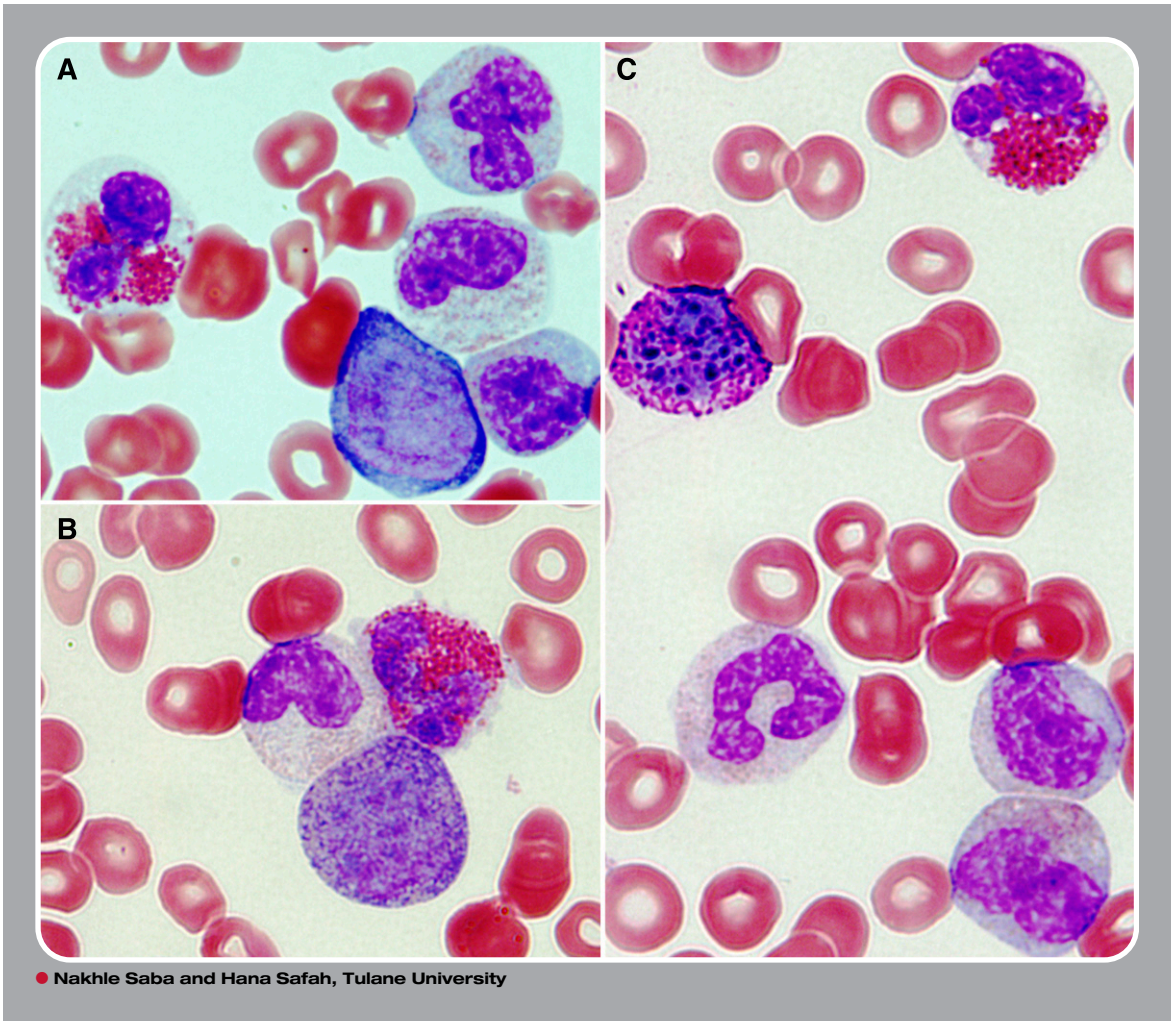
bloodwork
IMAGES IN HEMATOLOGY

A myeloproliferative neoplasm with translocation t(8;9)(p22;p24) involving *JAK2* gene



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A 35-year-old male presented with a several-weeks history of abdominal discomfort, early satiety, low-grade fever, night sweats, and weight loss. Physical examination revealed pale skin and a massive hepatosplenomegaly. A complete blood count showed severe anemia, thrombocytopenia, and marked leukocytosis (100 000/μL) with 10% blasts, eosinophilia (4000/μL), and basophilia (3000/μL). Peripheral smear demonstrated blasts (panel A), early myeloid precursors (panels A-C), eosinophilia (panels A-C), and basophilia (panel C). Bone marrow biopsy showed myeloid lineage hyperplasia. Cytogenetic analysis was significant for t(8;9)(p22;p24). Testing for *JAK2* mutation was negative, and FISH analysis failed to show *BCR-ABL1* or any abnormality involving *PDGFRA*, *PDGFRB*, or *FGFR1* genes. The patient was diagnosed with myeloproliferative neoplasm (MPN)-unclassified after excluding other hematologic neoplasms and causes of eosinophilia/basophilia.

Translocation t(8;9)(p22;p24) has been described to activate *JAK2*, resulting in a variety of diseases that surpass MPN to include cases of acute leukemia, lymphoma, and myelofibrosis. At the molecular level, t(8;9)(p22;p24) results in a new fusion gene product, putting the pericentriolar material 1 (*PCM1*) gene located on 8p22 in close proximity with the *JAK-2* gene located on 9p24. This results in the continuous activation of JAK2 tyrosine kinase. *PCM1-JAK2*–positive MPN is a rare disease characterized by an aggressive course and resistance to therapy.